


16	503.5	28.1	556	2	Q16GN5_AEDAE	Q16gn5 aedes aegyp	RT	cDNA.";
17	502.5	28.1	556	2	Q0IG10_AEDAE	Q0ig10 aedes aegyp	RL	Proc. Natl. Acad. Sci. U.S.A. 82:7289-7293(1985).
18	485	27.1	458	2	Q8MQ76_CAEEL	Q8mq76 caenorhabdi	RN	[2]
19	485	27.1	560	2	Q29BP0_DROPS	Q29bp0 drosophila	RP	NUCLEOTIDE SEQUENCE [MRNA] (ISOFORM SHORT).
20	453	25.3	551	2	Q8IMY3_DROME	Q8imy3 drosophila	RC	TISSUE=Hepatoma;
21	453	25.3	561	2	Q9VCJ4_DROME	Q9vcj4 drosophila	RX	MEDLINE=86085859; PubMed=3001061;
22	453	25.3	573	2	Q4V4F5_DROME	Q4v4f5 drosophila	RA	Tsuji S., Choudary P.V., Martin B.M., Winfield S., Barranger J.A.,
23	453	25.3	577	2	Q4V4J1_DROME	Q4v4j1 drosophila	RA	Ginns E.I.;
24	444	24.8	561	2	O16581_CAEEL	O16581 caenorhabdi	RT	"Nucleotide sequence of cDNA containing the complete coding sequence
25	441.5	24.7	523	2	O16580_CAEEL	O16580 caenorhabdi	RT	for human lysosomal glucocerebrosidase.";
26	441	24.6	524	2	Q611J0_CAEER	Q611j0 caenorhabdi	RL	J. Biol. Chem. 261:50-53(1986).
27	440.5	24.6	495	2	Q611J5_CAEER	Q611j5 caenorhabdi	RN	[3]
28	432.5	24.1	572	2	Q9AT27_PHYIN	Q9at27 phytophthor	RP	NUCLEOTIDE SEQUENCE [GENOMIC DNA].
29	421	23.5	424	2	Q4V4A3_DROME	Q4v4a3 drosophila	RC	TISSUE=Liver;
30	409	22.8	423	2	Q7PXX9_ANOGA	Q7pxx9 anopheles g	RX	MEDLINE=89122038; PubMed=2914709; DOI=10.1016/0888-7543(89)90319-4;
31	392.5	21.9	445	2	A4XMF5_CALSA	A4xmf5 caldicellul	RA	Horowitz M., Wilder S., Horowitz Z., Reiner O., Gelbart T.,
32	376.5	21.0	468	2	Q2AI75_9FIRM	Q2ai75 halothermot	RA	Beutler E.;
33	369.5	20.6	495	2	A3JON5_9FLAO	A3jon5 flavobacter	RT	"The human glucocerebrosidase gene and pseudogene: structure and
34	369	20.6	443	2	Q8R5Q0_THETN	Q8r5q0 thermoanaer	RT	evolution.";
35	345.5	19.3	494	2	A6EGQ6_9SPHI	A6egq6 pedobacter	RL	Genomics 4:87-96(1989).
36	340.5	19.0	445	2	Q1FLM6_9CLOT	Q1flm6 clostridium	RN	[4]
37	340.5	19.0	548	2	Q8PBP2_XANCP	Q8pbp2 xanthomonas	RP	NUCLEOTIDE SEQUENCE [GENOMIC DNA].
38	331	18.5	499	2	Q1VR53_9FLAO	Q1vr53 psychroflex	RC	TISSUE=Liver;
39	330.5	18.5	474	2	A2TWU2_9FLAO	A2twu2 polaribacte	RX	MEDLINE=92241881; PubMed=1572652; DOI=10.1016/0888-7543(92)90311-F;
40	328	18.3	489	2	Q3BWD0_XANCS	Q3bwd0 xanthomonas	RA	Beutler E., West C., Gelbart T.;
41	324	18.1	458	2	Q8PN97_XANAC	Q8pn97 xanthomonas	RT	"Polymorphisms in the human glucocerebrosidase gene.";
42	321.5	18.0	439	2	Q4URV7_XANCS	Q4urv7 xanthomonas	RL	Genomics 12:795-800(1992).
43	318.5	17.8	236	2	Q66IW2_XENLA	Q66iw2 xenopus lae	RN	[5]
44	318.5	17.8	487	2	A5FJMI_CYTJO	A5fjmi flavobacter	RP	NUCLEOTIDE SEQUENCE [MRNA] (ISOFORMS LONG AND 3), VARIANTS GD ARG-223;
45	313.5	17.5	490	2	Q1YUE9_9GAMM	Q1yue9 gamma prote	RP	GLY-230; PRO-235; ARG-241; ILE-252 AND ARG-364, AND VARIANTS GLY-310

ALIGNMENTS

RESULT 1

GLCM_HUMAN

ID GLCM_HUMAN  Reviewed; 536 AA.
AC P04062; Q16545; Q4VX22; Q6I9R6; Q9UMJ8;
DT 01-NOV-1986, integrated into UniProtKB/Swiss-Prot.
DT 09-NOV-2004, sequence version 3.
DT 21-AUG-2007, entry version 105.
DE Glucosylceramidase precursor (EC 3.2.1.45) (Beta-glucocerebrosidase)
DE (Acid beta-glucosidase) (D-glucosyl-N-acylsphingosine glucohydrolase)
DE (Alglucerase) (Imiglucerase).
GN Name=GBA; Synonyms=GC, GLUC;
OS Homo sapiens (Human).
OC Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
OC Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini;
OC Catarrhini; Hominidae; Homo.
OX NCBI_TaxID=9606;
RN [1]
RP NUCLEOTIDE SEQUENCE [MRNA] (ISOFORM SHORT).
RC TISSUE=Placenta;
RX MEDLINE=86042651; PubMed=3864160;
RA Sorge J., West C., Westwood B., Beutler E.;

RT "Molecular cloning and nucleotide sequence of human glucocerebrosidase

RT cDNA.";
RL Proc. Natl. Acad. Sci. U.S.A. 82:7289-7293(1985).
RN [2]
RP NUCLEOTIDE SEQUENCE [MRNA] (ISOFORM SHORT).
RC TISSUE=Hepatoma;
RX MEDLINE=86085859; PubMed=3001061;
RA Tsuji S., Choudary P.V., Martin B.M., Winfield S., Barranger J.A.,
RA Ginns E.I.;

RT "Nucleotide sequence of cDNA containing the complete coding sequence
for human lysosomal glucocerebrosidase.";
RL J. Biol. Chem. 261:50-53(1986).
RN [3]
RP NUCLEOTIDE SEQUENCE [GENOMIC DNA].
RC TISSUE=Liver;
RX MEDLINE=89122038; PubMed=2914709; DOI=10.1016/0888-7543(89)90319-4;
RA Horowitz M., Wilder S., Horowitz Z., Reiner O., Gelbart T.,
RA Beutler E.;

RT "The human glucocerebrosidase gene and pseudogene: structure and
evolution.";
RL Genomics 4:87-96(1989).
RN [4]
RP NUCLEOTIDE SEQUENCE [GENOMIC DNA].
RC TISSUE=Liver;
RX MEDLINE=92241881; PubMed=1572652; DOI=10.1016/0888-7543(92)90311-F;
RA Beutler E., West C., Gelbart T.;

RT "Polymorphisms in the human glucocerebrosidase gene.";
RL Genomics 12:795-800(1992).
RN [5]
RP NUCLEOTIDE SEQUENCE [MRNA] (ISOFORMS LONG AND 3), VARIANTS GD ARG-223;
RP GLY-230; PRO-235; ARG-241; ILE-252 AND ARG-364, AND VARIANTS GLY-310
RP AND HIS-368.
RX MEDLINE=94124033; PubMed=8294033; DOI=10.1016/0378-1119(93)90497-Q;
RA Imai K., Nakamura M., Yamada M., Asano A., Yokoyama S., Tsuji S.,
RA Ginns E.I.;

RT "A novel transcript from a pseudogene for human glucocerebrosidase in
RT non-Gaucher disease cells.";
RL Gene 136:365-368(1993).
RN [6]
RP NUCLEOTIDE SEQUENCE [GENOMIC DNA].
RX MEDLINE=97474796; PubMed=9331372;
RA Winfield S.L., Tayebi N., Martin B.M., Ginns E.I., Sidransky E.;

RT "Identification of three additional genes contiguous to the
RT glucocerebrosidase locus on chromosome 1q21: implications for Gaucher
RT disease.";
RL Genome Res. 7:1020-1026(1997).
RN [7]
RP NUCLEOTIDE SEQUENCE [LARGE SCALE GENOMIC DNA].
RX PubMed=16710414; DOI=10.1038/nature04727;
RA Gregory S.G., Barlow K.F., McLay K.E., Kaul R., Swarbreck D.,
RA Dunham A., Scott C.E., Howe K.L., Woodfine K., Spencer C.C.A.,
RA Jones M.C., Gillson C., Searle S., Zhou Y., Kokocinski F.,
RA McDonald L., Evans R., Phillips K., Atkinson A., Cooper R., Jones C.,
RA Hall R.E., Andrews T.D., Lloyd C., Ainscough R., Almeida J.P.,
RA Ambrose K.D., Anderson F., Andrew R.W., Ashwell R.I.S., Aubin K.,
RA Babbage A.K., Baggeley C.L., Bailey J., Beasley H., Bethel G.,
RA Bird C.P., Bray-Allen S., Brown J.Y., Brown A.J., Buckley D.,
RA Burton J., Bye J., Carder C., Chapman J.C., Clark S.Y., Clarke G.,

RA Clee C., Cobley V., Collier R.E., Corby N., Coville G.J., Davies J.,
 RA Deadman R., Dunn M., Earthrowl M., Ellington A.G., Errington H.,
 RA Frankish A., Frankland J., French L., Garner P., Garnett J., Gay L.,
 RA Ghorl M.R.J., Gibson R., Gilby L.M., Gillett W., Glithero R.J.,
 RA Grafham D.V., Griffiths C., Griffiths-Jones S., Grocock R.,
 RA Hammond S., Harrison E.S.I., Hart E., Haugen E., Heath P.D.,
 RA Holmes S., Holt K., Howden P.J., Hunt A.R., Hunt S.E., Hunter G.,
 RA Isherwood J., James R., Johnson C., Johnson D., Joy A., Kay M.,
 RA Kershaw J.K., Kibukawa M., Kimberley A.M., King A., Knights A.J.,
 RA Lad H., Laird G., Lawlor S., Leongamornlert D.A., Lloyd D.M.,
 RA Loveland J., Lovell J., Lush M.J., Lyne R., Martin S.,
 RA Mashreghi-Mohammadi M., Matthews L., Matthews N.S.W., McLaren S.,
 RA Milne S., Mistry S., Moore M.J.F., Nickerson T., O'Dell C.N.,
 RA Oliver K., Palmeiri A., Palmer S.A., Parker A., Patel D., Pearce A.V.,
 RA Peck A.I., Pelan S., Phelps K., Phillimore B.J., Plumb R., Rajan J.,
 RA Raymond C., Rouse G., Saenphimmachak C., Sehra H.K., Sheridan E.,
 RA Shownkeen R., Sims S., Skuce C.D., Smith M., Steward C.,
 RA Subramanian S., Sycamore N., Tracey A., Tromans A., Van Helmond Z.,
 RA Wall M., Wallis J.M., White S., Whitehead S.L., Wilkinson J.E.,
 RA Willey D.L., Williams H., Wilming L., Wray P.W., Wu Z., Coulson A.,
 RA Vaudin M., Sulston J.E., Durbin R.M., Hubbard T., Wooster R.,
 RA Dunham I., Carter N.P., McVean G., Ross M.T., Harrow J., Olson M.V.,
 RA Beck S., Rogers J., Bentley D.R.;
 RT "The DNA sequence and biological annotation of human chromosome 1.";
 RL Nature 441:315-321(2006).
 RN [8]
 RP NUCLEOTIDE SEQUENCE [LARGE SCALE MRNA] (ISOFORM LONG).
 RC TISSUE=Placenta;
 RX PubMed=15489334; DOI=10.1101/gr.2596504;
 RG The MGC Project Team;
 RT "The status, quality, and expansion of the NIH full-length cDNA
 RT project: the Mammalian Gene Collection (MGC).";
 RL Genome Res. 14:2121-2127(2004).
 RN [9]
 RP NUCLEOTIDE SEQUENCE [GENOMIC DNA / MRNA] OF 1-11.
 RX MEDLINE=88195776; PubMed=3359914;
 RA Reiner O., Wigderson M., Horowitz M.;
 RT "Structural analysis of the human glucocerebrosidase genes.";
 RL DNA 7:107-116(1988).
 RN [10]
 RP NUCLEOTIDE SEQUENCE [GENOMIC DNA] OF 1-45.
 RX MEDLINE=88074307; PubMed=3687939;
 RA Sorge J.A., West C., Kuhl W., Treger L., Beutler E.;
 RT "The human glucocerebrosidase gene has two functional ATG initiator
 RT codons.";
 RL Am. J. Hum. Genet. 41:1016-1024(1987).
 RN [11]
 RP PROTEIN SEQUENCE OF 40-44.
 RC TISSUE=Placenta;
 RA Martin B.M., Murray G.J., Coligan J.E., Raum M., Brady R.O.,
 RA Barranger J.A.;
 RT "Structural studies of human placental glucocerebrosidase.";
 RL Fed. Proc. 43:1869-1869(1984).
 RN [12]
 RP NUCLEOTIDE SEQUENCE [MRNA] OF 403-416.
 RX MEDLINE=85022513; PubMed=6091633;
 RA Ginns E.I., Choudary P.V., Martin B.M., Winfield S., Stubblefield B.,
 RA Mayor J., Merkle-Lehman D., Murray G.J., Bowers L.A., Barranger J.A.;
 RT "Isolation of cDNA clones for human beta-glucocerebrosidase using the
 RT lambda gt11 expression system.";
 RL Biochem. Biophys. Res. Commun. 123:574-580(1984).
 RN [13]
 RP NUCLEOTIDE SEQUENCE [GENOMIC DNA] OF 409-462, AND VARIANT GD SER-409.
 RC TISSUE=Skin;
 RX MEDLINE=88176943; PubMed=3353383;
 RA Tsuji S., Martin B.M., Barranger J.A., Stubblefield B.K.,
 RA LaMarca M.E., Ginns E.I.;
 RT "Genetic heterogeneity in type 1 Gaucher disease: multiple genotypes
 RT in Ashkenazic and non-Ashkenazic individuals.";
 RL Proc. Natl. Acad. Sci. U.S.A. 85:2349-2352(1988).
 RN [14]
 RP PROTEIN SEQUENCE OF 469-520.
 RC TISSUE=Placenta;
 RX MEDLINE=86149363; PubMed=3456607;
 RA Dinur T., Osiecki K.M., Legler G., Gatt S., Desnick R.J.,
 RA Grabowski G.A.;
 RT "Human acid beta-glucosidase: isolation and amino acid sequence of a
 RT peptide containing the catalytic site.";
 RL Proc. Natl. Acad. Sci. U.S.A. 83:1660-1664(1986).
 RN [15]
 RP MASS SPECTROMETRY, MUTAGENESIS OF GLU-379, AND ACTIVE SITE.
 RX PubMed=7908905;
 RA Miao S., McCarter J.D., Grace M.E., Grabowski G.A., Aebersold R.,
 RA Withers S.G.;
 RT "Identification of Glu340 as the active-site nucleophile in human
 RT glucocerebrosidase by use of electrospray tandem mass spectrometry.";
 RL J. Biol. Chem. 269:10975-10978(1994).
 RN [16]
 RP INTERACTION WITH SAPOSIN-C AND MEMBRANES CONTAINING ANIONIC
 RP PHOSPHOLIPIDS.
 RX PubMed=10781797; DOI=10.1016/S0014-5793(00)01417-4;
 RA Salvioli R., Tatti M., Ciaffoni F., Vaccaro A.M.;
 RT "Further studies on the reconstitution of glucosylceramidase activity
 RT by Sap C and anionic phospholipids.";
 RL FEBS Lett. 472:17-21(2000).
 RN [17]
 RP GLYCOSYLATION AT ASN-98; ASN-185 AND ASN-309.
 RX MEDLINE=22660472; PubMed=12754519; DOI=10.1038/nbt827;
 RA Zhang H., Li X.-J., Martin D.B., Aebersold R.;
 RT "Identification and quantification of N-linked glycoproteins using
 RT hydrazide chemistry, stable isotope labeling and mass spectrometry.";
 RL Nat. Biotechnol. 21:660-666(2003).
 RN [18]
 RP X-RAY CRYSTALLOGRAPHY (2.0 ANGSTROMS) OF 40-536, GLYCOSYLATION AT
 RP ASN-58, AND DISULFIDE BONDS.
 RX PubMed=12792654; DOI=10.1038/sj.embor.embor873;
 RA Dvir H., Harel M., McCarthy A.A., Toker L., Silman I., Futerman A.H.,
 RA Sussman J.L.;
 RT "X-ray structure of human acid-beta-glucosidase, the defective enzyme
 RT in Gaucher disease.";
 RL EMBO Rep. 4:704-709(2003).
 RN [19]
 RP X-RAY CRYSTALLOGRAPHY (2.4 ANGSTROMS) OF 40-536 IN COMPLEX WITH
 RP SYNTHETIC INHIBITOR, AND ACTIVE SITE.

RX PubMed=15817452; DOI=10.1074/jbc.M502799200;
 RA Premkumar L., Sawkar A.R., Boldin-Adamsky S., Toker L., Silman I.,
 RA Kelly J.W., Futerman A.H., Sussman J.L.;
 RT "X-ray structure of human acid-beta-glucosidase covalently bound to
 RT conduritol-B-epoxide. Implications for Gaucher disease.";
 RL J. Biol. Chem. 280:23815-23819(2005).
 RN [20]
 RP X-RAY CRYSTALLOGRAPHY (2.5 ANGSTROMS) OF 40-536, CHARACTERIZATION OF
 RP GD VARIANTS SER-55; GLN-87; ASN-118; GLN-159; LEU-161; VAL-162;

Query Match 94.2%; Score 1687; DB 1; Length 536;
 Best Local Similarity 67.0%; Pred. No. 8.6e-137;
 Matches 333; Conservative 1; Mismatches 1; Indels 162; Gaps 1;

Qy 1 ARPCIPKSFSGYSVVCVNCNATYCDSPDPTFPALGTFSTRYESTRSGRRMELSMGPIQANH 60
 |
 Db 40 ARPCIPKSFSGYSVVCVNCNATYCDSPDPTFPALGTFSTRYESTRSGRRMELSMGPIQANH 99
 Qy 61 TGTGLLLTLQPEQKFKVKVGGAMTDAALNIALSPPAQNLLKSYFSEEG----- 113
 |
 Db 100 TGTGLLLTLQPEQKFKVKVGGAMTDAALNIALSPPAQNLLKSYFSEEGIGYNIIR 159
 Qy 114 ----- 113
 Db 160 VPMASCDFESIRTYTYADTPDDFQLHNFSLPEEDTKLKIPLIHRALQLAQRPVSLASPWT 219
 Qy 114 ----- 113
 Db 220 SPTWLKTINGAVNGKSLKQPGDIYHQTWARYFVKFLDAYAEHKLQFMAVTAENEPSAGL 279
 Qy 114 -----VRLMLNDQRLLLPHWAKVVLTDPE 138
 |
 Db 280 LSGYPFQCLGFTPEHQDFIARDLGPGLANSTHNVRLMLDDQRLLPHWAKVVLTDPE 339
 Qy 139 AAKYVHGI AVHWYLDLFLAPAKATLGETHRLFPNTMLFASEACVGSKFWEQSVRLGSWDRG 198
 |
 Db 340 AAKYVHGI AVHWYLDLFLAPAKATLGETHRLFPNTMLFASEACVGSKFWEQSVRLGSWDRG 399
 Qy 199 MQYSHSIITNLLYHVVGWTDWNLALNPEGGPNWRNFVDSPIIVDITKDTFYKQPMFYHL 258
 |
 Db 400 MQYSHSIITNLLYHVVGWTDWNLALNPEGGPNWRNFVDSPIIVDITKDTFYKQPMFYHL 459
 Qy 259 GHFSKFIPEGSQRVGLVASQKNDLDAVALMHPDGSAAVVVLMNRSSKDVPLTIKDPVAVGFL 318
 |
 Db 460 GHFSKFIPEGSQRVGLVASQKNDLDAVALMHPDGSAAVVVLMNRSSKDVPLTIKDPVAVGFL 519
 Qy 319 ETISPGYSIHLYLWHRQ 335
 |
 Db 520 ETISPGYSIHLYLWRRQ 536

RESULT 2
 GLCM_PANTR
 ID GLCM_PANTR Reviewed; 536 AA.
 AC Q9BTD0;
 DT 21-NOV-2003, integrated into UniProtKB/Swiss-Prot.
 DT 01-JUN-2001, sequence version 1.